AMINO ACID DISORDERS

What is it?

Amino acid disorders are a group of inherited metabolic conditions that lead to a buildup of amino acids and/or bi-products of amino acid metabolism in the blood causes severe medical complications

Babies born with one of these disorders cannot breakdown certain waste products from their blood, such as amino acids or ammonia. This can lead to problems with the eyes, skin or general development, liver failure, coma or death if untreated. Treatment can range from special diets to liver transplantation and special medications. A baby with an amino acid disorder must have regular medical care by an experienced physician. The Newborn Bloodspot Screening Program will screen for:

- Argininosuccinic aciduria (ASA)
- Citrullinemia (ASS)
- Homocystinuria (HCU)
- Hypermethioninemia
- Maple syrup urine disease (MSUD)
- Tyrosinemia, type II

How do you get it?

All amino acid disorders are inherited in an autosomal recessive pattern. As and autosomal recessive disorder, the parents of a child with one of these conditions are unaffected, healthy carrier of the condition, and have normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene and the result amino acid defect. Carrier parents have a 50 percent chance of having an unaffected, non-carrier child. These risks would hold true of each pregnancy. All siblings of infants diagnosed with an organic acid disorder should be tested.

Many amino acid disorders present in the neonatal period. Typically, an affected newborn appears normal for the first days of life but depending upon the disorder may then develop lethargy, poor feeding, failure to thrive, respiratory distress, developmental delay and mental retardation if left untreated and in some cases an unusual odor.

How common is it?

Phenylketonuria (PKU) is the most common of the amino acid disorders with an incidence of approximately 1 in 15,000 births.

Estimates vary widely for the incidence of each amino acid disorder and for many the actual incidence is not yet known, however most of these disorders are quite rare.

How is it treated?

For many amino acid disorders, early diagnosis and treatment can change the outcome of the disorder. Treatment depends upon the specific amino acid disorder.

If the child needs additional testing or diagnostic evaluation it is important that you follow through. Treatment is life long and compliance with dietary management is imperative to your child's health, growth and development.

Infants and children with an amino acid disorder should have regular follow-up appointments with a metabolic specialist. If your infant shows early signs of the condition, such as seizures or lethargy, immediately seek medical care. The metabolic specialist and the primary care provider should develop a medical plan for these acute episodes.

Long-term management, monitoring and compliance with treatment recommendations are essential to your child's well being. A multi-disciplinary approach, including the following specialties is recommended: pediatrics, genetics and nutrition.

Where can I get services?

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

Related Links

Centers for Disease Control and Prevention www.cdc.gov

Medline Plus (National Library of Medicine and the National Institutes of Health) www.medlineplus.gov

National Coalition for PKU and Allied Disorders www.pku-allieddisorders.org

National Library of Medicine (NLM) Pub Med www.ncbi.nlm.nih.gov

American College of Medical Genetics www.acmg.net/